



CP gene

ceruloplasmin

Normal Function

The *CP* gene provides instructions for making a protein called ceruloplasmin. Ceruloplasmin helps move iron from the organs and tissues of the body into the blood. This protein prepares iron for incorporation into a molecule called transferrin, which transports the iron to red blood cells.

There are two forms of ceruloplasmin. One form, serum ceruloplasmin, is made primarily in the liver. It is involved in transporting iron from most of the body, but is unable to enter the brain. The other form of ceruloplasmin, called the glycosylphosphatidylinositol (GPI)-anchored form, is important for processing iron in the brain and releasing it from brain tissue. This form of ceruloplasmin is made in nervous system cells called glia, which protect and maintain nerve cells (neurons).

Health Conditions Related to Genetic Changes

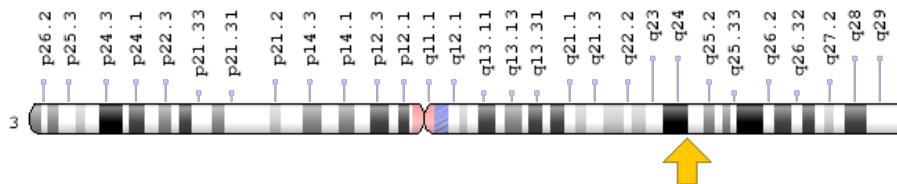
aceruloplasminemia

Approximately 40 mutations in the *CP* gene that cause aceruloplasminemia have been identified. Some of these mutations substitute one protein building block (amino acid) for another amino acid in the ceruloplasmin protein, resulting in an unstable protein that quickly breaks down (degrades). Other mutations result in the production of an abnormally short, nonfunctional version of the protein or prevent the protein from being secreted by the cells in which it is made. Absence of functional ceruloplasmin results in iron transport problems that lead to the iron accumulation, neurological dysfunction, and other health problems seen in aceruloplasminemia.

Chromosomal Location

Cytogenetic Location: 3q24-q25.1, which is the long (q) arm of chromosome 3 between positions 24 and 25.1

Molecular Location: base pairs 149,162,410 to 149,222,050 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CERU_HUMAN
- ceruloplasmin (ferroxidase)
- CP-2
- ferroxidase

Additional Information & Resources

GeneReviews

- Aceruloplasminemia
<https://www.ncbi.nlm.nih.gov/books/NBK1493>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CP%5BTIAB%5D%29+OR+%28ceruloplasmin%5BTIAB%5D%29%29+AND+%28%28alpha%282%29-ceruloplasmin%5BMAJR%5D%29+OR+%28ferroxidase%5BMAJR%5D%29+OR+%28ceruloplasmin%5BMAJR%5D%29+OR+%28ceruloplasmin+oxidase%5BMAJR%5D%29+OR+%28ceruloplasmin+ferroxidase%5BMAJR%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- CERULOPLASMIN
<http://omim.org/entry/117700>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CP.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CP%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2295
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1356>
- UniProt
<http://www.uniprot.org/uniprot/P00450>

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Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/CP>

Reviewed: October 2013
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services